STATE OF WASHINGTON
WASHINGTON STATE BOARD OF HEALTH
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October 12, 2005

TO: Washington State Board of Health Members

FROM: Dr. Kim Marie Thorburn, Board Chair

SUBJECT: NEWBORN CYSTIC FIBROSIS SCREENING AND THE ACMG REPORT

Background and Summary

In 2002 Dr. Thomas Locke and Dr. Maxine Hayes concluded their work with a Newborn Screening Advisory Committee, which had been asked to: (1) develop criteria to use when considering new disorders for the mandatory screening panel and (2) consider whether any additional disorders should be added to the panel.

In 2003 the Board adopted five criteria to consider when evaluating new disorders, and it added to the screening panel five new disorders that had been evaluated using the new criteria. The Advisory Committee did not recommend adding cystic fibrosis at that time but asked that the Board re-evaluate this decision in two years as more science became available.

At its December 2004 meeting, the Board asked the Department of Health (DOH) to begin the re-evaluation process. In April 2005 a panel of technical experts reviewed the new science that was available and recommended that a larger advisory panel proceed with a full evaluation of cystic fibrosis using the five criteria adopted by the Board in 2003. The larger advisory committee met in July 2005 to further review cystic fibrosis against the five criteria and make recommendations to the Board. These recommendations are included in your packet (under Tab 6 in the *Review of Criteria for Adding Cystic Fibrosis to NBS Program*).

Medical and technological advances in recent years have made it feasible to screen newborns for a larger number of disorders. Many disorders can be detected using the same dried blood specimen that is routinely collected to test infants in Washington State. In 2004, the United States Department of Health and Human Services' Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children accepted a report commissioned from the American College of Medical Genetics (ACMG). This report recommends 29 disorders for newborn screening. Eighteen of these twenty-nine disorders (including hearing loss and cystic fibrosis) are currently not screened for in Washington State.

Today, I have invited Dr. Thomas Locke, former Board Chair; Dr. Maxine Hayes, State Health Officer; and Mike Glass, Director of the Washington State Newborn Screening Program, to talk about the recommendations from the cystic fibrosis advisory committee. I have also asked DOH staff to update the Board on the Newborn Screening Annual Report findings and Specialty Clinic fees. According to WAC 246-650-040, DOH is required to provide annual reports to the Board on the following information concerning tests: (1) the costs of tests as charged by DOH; (2) the results of each category of tests, by county of birth and ethnic group, as reported on the newborn screening form and, if available, birth certificates; and (3) follow-up procedures and the results of such follow-up procedures.

Finally, I have invited DOH to present its review of the ACMG report. In December 2005, the Board asked DOH to determine if there are effective interventions for the 16 conditions that have not yet been evaluated for inclusion in Washington's screening battery, and also to provide some sense of the associated costs; Washington's capacity to detect and treat the disorders; and the number of newborns in Washington who could potentially be identified with the conditions. DOH has reviewed the report as requested and will present its findings today (see Tab 6 *Synopsis: National Recommendations*).

Recommended Board Action

Motion 1:

The Board requests that DOH continue the rule making process that would add cystic fibrosis to the list of preventable heritable disorders leading to developmental disabilities or physical defects in Chapter 246-650 WAC.

Motion 2:

The Board will work with the Department of Health to begin a process to review the conditions set forth in the American College of Medical Genetics' report that are currently not under consideration by the Board or included in Chapter 246-650 WAC. The conditions will be reviewed against the five Board-approved criteria for adding disorders to the newborn screening program and recommendations will be made regarding which, if any, the Board should consider adopting in rule.

Discussion

Cystic Fibrosis

The rule: Washington's newborn screening law (Chapter 70.83 RCW—Phenylketonuria and Other Preventable Heritable Disorders) states that "the policy of the state of Washington to make every effort to detect as early as feasible and to prevent where possible phenylketonuria and other preventable heritable disorders leading to developmental disabilities or physical defects." The statute authorizes the Board to determine which disorders in addition to phenylketonuria (PKU) are to be included in newborn screening required by the state. The statute also delegates authority toDOH to require that all newborns receive screenings for the detection of the disorders that are defined by the Board before they are discharged from the hospital.

In Chapter 246-650 WAC, the Board has identified eight "preventable heritable disorders leading to developmental disabilities or physical defects" in addition to PKU. Currently, Washington State requires screening for PKU, congenital hypothyroidism, congenital adrenal hyperplasia,

hemoglobinopathies such as sickle cell disease (which includes sickle cell anemia, Hb S/Beta-thalassemia, and Hb S/C disease), biotinidase deficiency, galactosemia, homocystinuria, medium chain acyl Co-A dehydrogenase deficiency (MCADD), and Maple Syrup Urine Disease (MSUD). DOH tests for these disorders at the State Public Health Laboratory, an activity supported by a charge collected through the hospital or other birth facility.

The Process: Dr. Hayes and Dr. Locke co-chaired the Newborn Screening Advisory Committee, which concluded its work in 2002. In 2003 the Newborn Screening Advisory Committee made a number of recommendations to the Board. As a result, the Board adopted five criteria for evaluating any additional mandated newborn screenings and accepted a recommendation to begin screening for five new disorders. The five criteria include: (1) prevention potential and medical rationale, (2) treatment available, (3) public health rationale, (4) available technology, and (5) cost-benefit/cost-effectiveness. Any disorder that met the first four criteria, the committee recommended, should then be evaluated using the fifth criteria. Although the committee did not recommend adding cystic fibrosis to the list at that time, it strongly encouraged the Board to re-evaluate this decision in two years when the results of several additional studies on early cystic fibrosis treatment would be available. Since 2002, several studies have shed new light on therapeutic interventions and cost benefit/cost effectiveness of cystic fibrosis treatment.

At its meeting in December 2004, the Board approved a motion to work with DOH to convene a panel of technical experts to review new information available on the benefits of newborn screening for cystic fibrosis and make a preliminary determination whether this condition meets criteria established for newborn screening tests in Washington. A technical review committee of seven experts in public health and cystic fibrosis met on April 9, 2005. The committee's charge was to review current scientific and medical evidence regarding newborn screening for cystic fibrosis against the Board's five criteria for adding disorders to the state's mandatory screening program. The committee was asked if the evidence was sufficiently compelling to justify convening a broadly representative advisory committee to review all of the issues and make a formal recommendation to the Board as to whether cystic fibrosis should be added to the state's mandatory requirements. The technical review committee unanimously concluded that the research evidence is consistent with the criteria and that a larger advisory committee should be convened.

The larger advisory committee met in July 2005 to further review cystic fibrosis against the five criteria and make recommendations to the Board. The broader advisory committees' votes were unanimous on the first four criteria. While the majority (or nine of the fourteen committee members) felt the fifth criterion (cost-benefit/cost-effectiveness) was met, three of the committee members did not agree and there were also two abstentions. Several methodological issues arose that were subsequently addressed in consultation with a national expert. The revised analysis concludes that an estimated $5.40 in benefit will be realized for each dollar spent on screening-related costs.

The advisory committee also felt that careful implementation would be necessary to achieve the desired benefits, and made four implementation recommendations. These accompany the committee's scoring on the five criteria.